



“III European Days of Albinism Milan”

Milan, 7-8 April 2016

Thursday, 7 April 2016

08:00 -08:30 Registration

08:30-09:00 General welcome

Chair: *Tronconi E, Marocchi A, Gesu G, Representative of the Region, Masto R, Paschetta L*

09:00-10:00 Scientific Session I

Clinical Definition of Albinism

Chair: *Gottlob I, Piozzi E*

- Expanding the phenotype of albinism? - **Barbara Käsmann-Kellner** (20 min)
- Phenotypic spectrum in FHONDA and albinism – **Maria Van Genderen** (20 min)
- *Discussion* (20 min)

10:00 -11.30 Scientific Session II

Clinical and Molecular Diagnosis of Albinism

Chair: *Grønskov K, Scalinci S*

- Postnatal retinal development in albinism as compared to healthy controls – **Irene Gottlob** - (20 min)
- Visual cortex in albinism - anatomy and function – **Michael Hoffmann** (20 min)
- Contribution of Electrofunctional Examinations in Albinism Diagnosis – **Alma Patrizia Tormene** (15 min)
- Molecular diagnostic approaches: Targeted sequencing, exome, array-CGH? - **Benoit Arveiler** (20 min)
- *Discussion* (15 min)

11:30-12:00 coffee break

12:00-13:30 Scientific Session III
European Diagnostic Experiences

Chair: Del Longo A, Taïeb A, Primignani P

- Clinical and molecular experience in Denmark– **Karen Grønskov** (20 min)
- Clinical and molecular experience in Italy – **Roberta Terrana, Lucia Mauri** (15 min)
- Albino Day in Madrid- **Marta Cortón** (15 min)
- Albino Day in Bordeaux – **Fanny Maurice Picard** (15 min)
- Discussion (20 min)

13:30-14:40 Lunch break

14:40- 16:00 Scientific Session IV
Pathogenesis of Albinism

Chair: Montoliu L, Schiaffino MV,

- Understanding visual defects in albinism using mouse and cellular models - **Alexandra Rebsam** (20 min)
- Syndromic albinism disorders affecting membrane trafficking during melanosome formation- **Michael Marks** (20 min)
- Melanosomal ion channels critical for pigmentation – **Elena Oancea** (20 min)
- Discussion (20 min)

16:00-16.30 Coffee/Tea break

16:30-18:10 Scientific Session V:
Beyond Eyes

Chair: Arveiler B., Penco S.

- Oculocutaneous albinism in East and Southern Africa - **Patricia Lund** (20 min)
- Skin photoprotection issues in albinism - **Alain Taïeb** (20 min)
- Dermatological characterization in Italian albinos –**Marco Antonio Negri** (20 min)
- Audiological deficits in albinos: correlation with genetic/clinical phenotype – **Giuseppe Mingoia** (20 min)
- Discussion (20 min)

Friday, 8 April 2016

8.30-10:00 Scientific Session VI

Technical Support and Therapeutic Frontiers

Chair: Käsmann-Kellner B, Del Longo A

- Visual management training in low vision patients - **Flavia Fabiani** (15 min)
- Pharmacological therapy – **Brian Brooks** (20 min)
- Defining safety and efficacy end-points for OCAI gene therapy-based clinical studies– **Enrico Surace** (15 min)
- New animal models of albinism generated with CRISPR tools – **Luis Montoliu** (20 min)
- Discussion (20 min)

10.00 – 10.30 Final Scientific Discussion:

Chairs: Schiaffino MV, Marocchi A, Gottlob I, Montoliu L

- Discussion (30 min)

Plenary final session: scientists and associations together

- 10.30 -11:00 Summary of scientific presentations (*Käsmann-Kellner B, Patrosso MC*)
- 11:00 - 11.20 Summary of psycho pedagogical presentations (20 min)
- 11.20 -11.50 Summary of association's presentations (30 min)
- 11.50- 12.00 Kid for kindness presentation (10 min)
- 12.00- 12.10 Dermatological Prevention presentation (10 min)
- 12.10-13.30 Global discussion/Presentation of leaflet/book on albinism for the general public/Proposal for the next meeting.